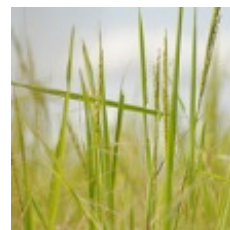
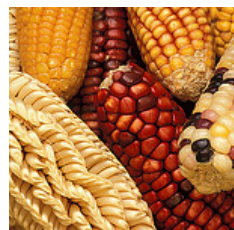




Variation at NCBI: Resources, Tools and Submissions

Jennifer M. Lee

PAG XXIII January 10-14, 2014



Poster 1109 – dbSNP and dbVar

NCBI Genomic Variation Databases

Database	Content	Public (Year)
dbSNP	Simple Genetic Variations	1998
dbMHC	DNA and clinical data related to the human Major Histocompatibility Complex	2002
dbRBC	DNA and clinical data related to Red Blood Cells	2002
dbGaP	Genotypes and Phenotypes (open and controlled)	2006
dbVar	CNV and Complex Structural Variations	2009
GTR	Genetic Testing Registry	2010
ClinVar	Aggregates clinical variation and its relationship to human health	2014



New Variation Submission Portal

<http://submit.ncbi.nlm.nih.gov/variation>

NCBI

Variation Submission Portal

[New organization](#) [Home](#) [Contact](#) [Help](#) [Jennifer Lee](#) [Log out](#)

Your organizations

[GeneDx](#) [Edit](#) [Request deletion](#)

Organization information

Department: GeneDx
Institution: GeneDx
Address: 207 Perry Parkway, Gaithersburg, Maryland, United States 20877
Status: Processed-ok
Organization ID: 26957

Submitted data - 2 submissions [Upload submission](#)

Filter Submissions by Status or other fields:

Submission ID	Submission name	Status	Submitter name	Created date	
SUB779344	KAT6A_limited	created	Lisa Vincent	Dec 22, 2014	Edit Delete
SUB737145	GDX_Cancer_10_28_2014	processing	Lisa Vincent	Nov 05, 2014	

Register your organization once and any information about the organization required for submission is automatically added to the submission.

There is now one location to submit variation data to NCBI. Submissions can be tracked.

New (SUB123456)

Unfinished at the Organization information step

[Organization information](#) [Submitter information](#) [Overview](#)

Organization Information

* Organization name [?](#)

Institution name [?](#)

Department name [?](#)

Street address [?](#)

* City [?](#)

State or Province [?](#)

* Country or Region [?](#)

Postal code [?](#)

Organization URL [?](#)

Base URL for variant links [?](#)

☒ Report and acknowledge my organization in public reports from ClinVar [?](#)

[Continue](#)

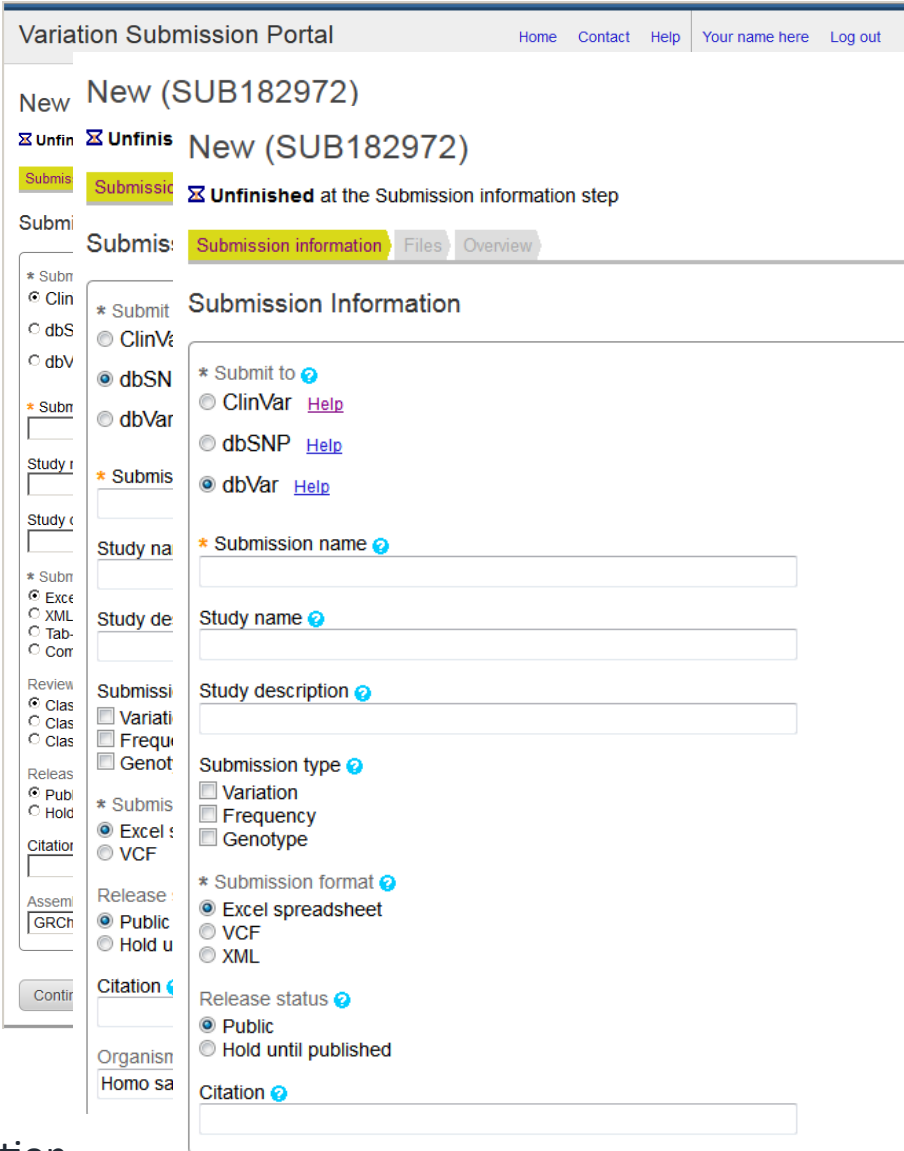
If you have the id GTR or ClinVar uses for your organization, [use this link to display what is stored.](#)



National Center for Biotechnology Information

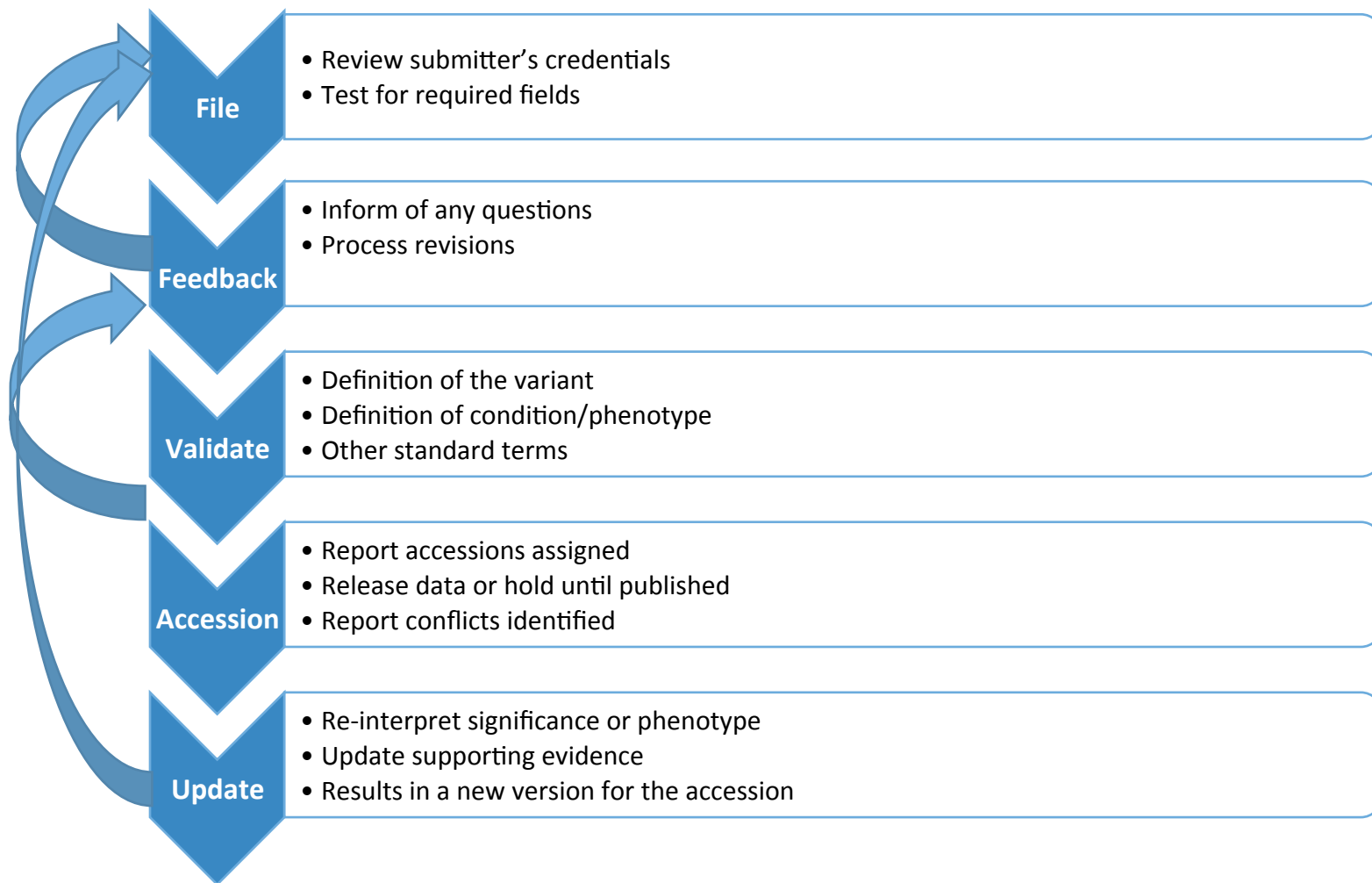
Variation submission portal

- Select the resource to which you would like to submit data.
- Provide basic information about your submission
- Upload the submission file.
- Accepts vcf format for dbSNP and dbVar



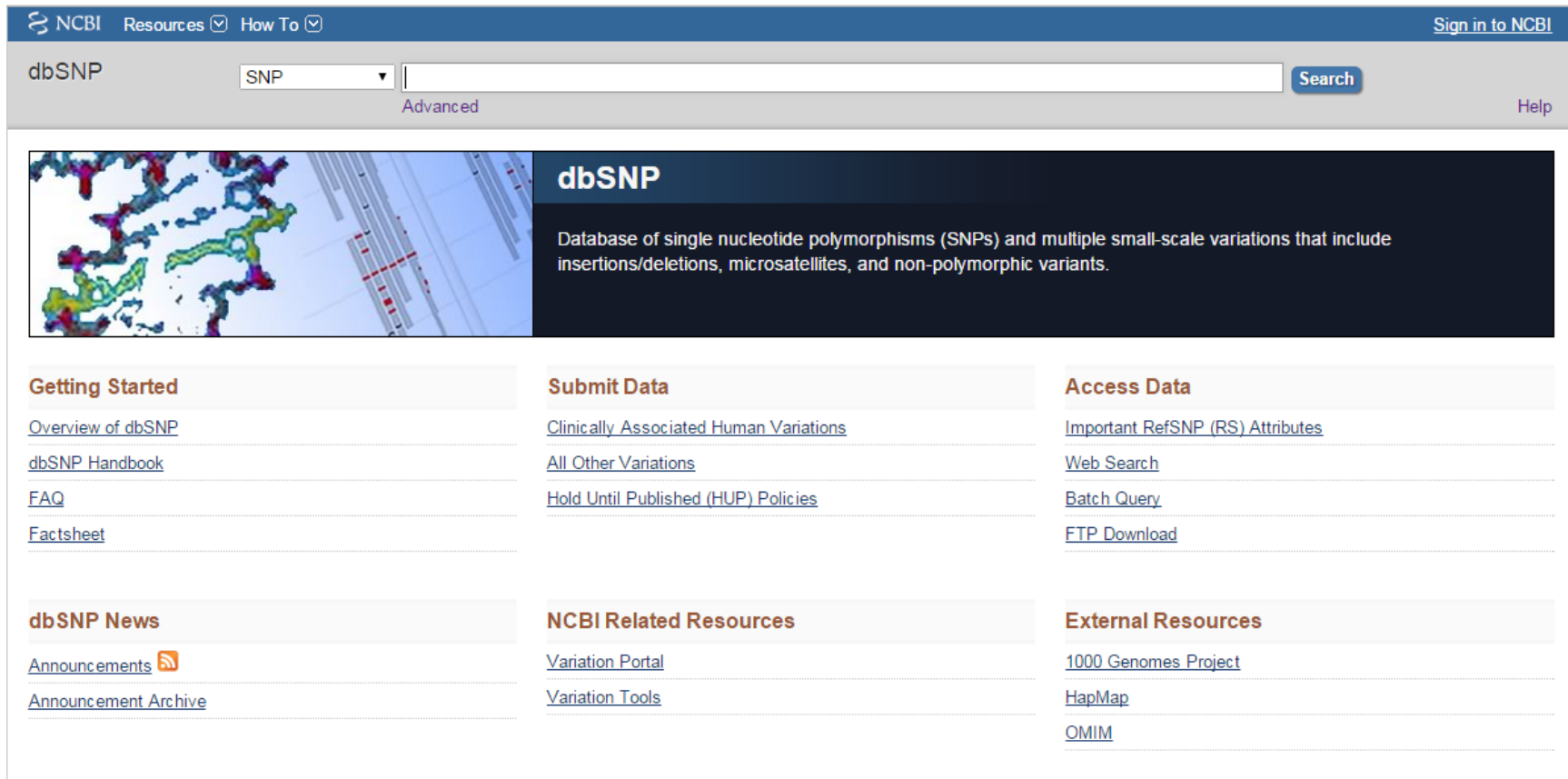
The screenshot displays the 'Variation Submission Portal' interface. At the top, there is a navigation bar with links for 'Home', 'Contact', 'Help', 'Your name here', and 'Log out'. The main heading is 'New (SUB182972)'. Below this, there are tabs for 'Unfin', 'Unfinis', and 'New (SUB182972)'. A status message indicates 'Unfinished at the Submission information step'. The 'Submission information' tab is active, showing a form with various fields and options. On the left side, there is a sidebar with a tree view containing 'Subn', 'Clin', 'dbS', 'dbV', 'Subn', 'Study', 'Study', 'Subn', 'Excel', 'XML', 'Tab', 'Corr', 'Review', 'Clas', 'Clas', 'Clas', 'Releas', 'Publ', 'Hold', 'Citation', 'Assem', 'GRCh', and 'Contir'. The main form area includes sections for 'Submit to' (with radio buttons for ClinVar, dbSNP, and dbVar), 'Submission name', 'Study name', 'Study description', 'Submission type' (with checkboxes for Variation, Frequency, and Genotype), 'Submission format' (with radio buttons for Excel spreadsheet, VCF, and XML), 'Release status' (with radio buttons for Public and Hold until published), and 'Citation'. The 'Organism' field is set to 'Homo sa'.

Overview of submission processing



dbSNP - Primary archive for common and rare short sequence variation (somatic and germline)

<http://www.ncbi.nlm.nih.gov/snp>



The screenshot shows the NCBI dbSNP website. At the top is a navigation bar with the NCBI logo, links for 'Resources' and 'How To', and a 'Sign in to NCBI' link. Below this is a search bar with 'dbSNP' in the dropdown, a search input field, and a 'Search' button. A 'Help' link is also present. The main content area features a large image of a DNA microarray on the left and a dark blue banner on the right with the text 'dbSNP Database of single nucleotide polymorphisms (SNPs) and multiple small-scale variations that include insertions/deletions, microsatellites, and non-polymorphic variants.' Below the banner are three columns of links. The first column, 'Getting Started', includes links for 'Overview of dbSNP', 'dbSNP Handbook', 'FAQ', and 'Factsheet'. The second column, 'Submit Data', includes links for 'Clinically Associated Human Variations', 'All Other Variations', and 'Hold Until Published (HUP) Policies'. The third column, 'Access Data', includes links for 'Important RefSNP (RS) Attributes', 'Web Search', 'Batch Query', and 'FTP Download'. Below these columns are three more sections: 'dbSNP News' with links for 'Announcements' and 'Announcement Archive'; 'NCBI Related Resources' with links for 'Variation Portal' and 'Variation Tools'; and 'External Resources' with links for '1000 Genomes Project', 'HapMap', and 'OMIM'.

NCBI Resources How To Sign in to NCBI

dbSNP SNP Search Help

dbSNP

Database of single nucleotide polymorphisms (SNPs) and multiple small-scale variations that include insertions/deletions, microsatellites, and non-polymorphic variants.

Getting Started

- [Overview of dbSNP](#)
- [dbSNP Handbook](#)
- [FAQ](#)
- [Factsheet](#)

Submit Data

- [Clinically Associated Human Variations](#)
- [All Other Variations](#)
- [Hold Until Published \(HUP\) Policies](#)

Access Data

- [Important RefSNP \(RS\) Attributes](#)
- [Web Search](#)
- [Batch Query](#)
- [FTP Download](#)

dbSNP News

- [Announcements](#)
- [Announcement Archive](#)

NCBI Related Resources

- [Variation Portal](#)
- [Variation Tools](#)

External Resources

- [1000 Genomes Project](#)
- [HapMap](#)
- [OMIM](#)



dbSNP Variation Classes

- Types
 - SNV (common and rare)
 - MNP
 - Short INDEL
 - MicroSAT
 - STR
- Allele Origin
 - Germline
 - Somatic



Content dbSNP

350 Organisms
1.3B Submitted SNP “SS Accessions”
0.6B Reference SNP “RS Accessions”

TAXID	ORGANISM	SS COUNT	RS COUNT
9913	<i>Bos taurus</i>	234957220	85027819
9940	<i>Ovis aries</i>	100819215	54004457
9823	<i>Sus scrofa</i>	82712826	28702828
9925	<i>Capra hircus</i>	55076618	37166653
9031	<i>Gallus gallus</i>	41509399	9526068
3847	<i>Glycine max</i>	32069807	13087062
469796	<i>Ovis orientalis</i>	29547990	29263208
4577	<i>Zea mays</i>	13784397	7200277
4530	<i>Oryza sativa</i>	13271151	10966784
9796	<i>Equus caballus</i>	5572544	4974054
	more....		



dbSNP

SNP

bovine

Save search Advanced

Help

[Show additional filters](#)

[Clear all](#)

Organism

Bos taurus

More ...

Variation Class

in del

snp

Annotation

Cited in PubMed

PubMed

nucleotide

structure

Function Class

3' splice site

3' utr

5' splice site

5' utr

coding synonymous

frame shift

intron

missense

nonsense

stop gained

Global MAF

Custom range...

Validation Status

by-frequency

no-info

[Clear all](#)

[Show additional filters](#)

Display Settings: ☒ Summary, 20 per page, Sorted by SNP_ID

Send to: ☒ Filter your results:

Results: 1 to 20 of 874117

<< First < Prev Page 1 of 43706 Next > Last >>

Filters activated: missense. Clear all to show 73439640 items.

Manage Filters

☐ rs8193049 [Bos taurus]

1.

GACTTCCCATTGGACATCTCAAAA[A/C]CTTGAAAGAGCTTAATGTGGCTCAC

Chromosome: 8:108837116

Gene: TLR4 (GeneView)

Functional Consequence: missense

Validated: no info

[PubMed](#) [Protein3D](#)

☐ rs8193055 [Bos taurus]

2.

CCAAGCCTTCAGTATCTAGATCTCA[A/G]AAGAAATCACTTGAGTTTCAAGGGT

Chromosome: 8:108837806

Gene: TLR4 (GeneView)

Functional Consequence: missense

Validated: no info

[PubMed](#) [Protein3D](#)

☐ rs17871971 [Bos taurus]

3.

GTGGTGTTTCCAAAGGGCTGGTCAC[A/G]GTCTCAGGCTTGGGTGTGGCCTGG

Chromosome: 5:104223951

Gene: NOP2 (GeneView)

Functional Consequence: missense

Validated: no info

[PubMed](#)

☐ rs55617272 [Bos taurus]

4.

AAATAAAATCTCAAAAATTCAGAGT[A/G]GTGCTTTTCTTGTTGGGGCACCT

Chromosome: 27:15240722

Gene: TLR3 (GeneView)

Functional Consequence: missense

Validated: no info

[PubMed](#) [Protein3D](#)

Find related data

Database: Select

Find items

Search details

"Bos taurus"[Organism] AND missense[Function_Class]

Search

See more...

Recent activity

Turn Off Clear

bovine AND (missense[Function_Class]) (874117) SNP

bovine (73439640) SNP

Diversity and evolution of 11 innate immune genes in Bos taurus taurus and Bos t. PubMed

Pubmed (SNP Cited) for SNP (Select 8193049) (1) PubMed

Genomic characteristics of cattle copy number variations. PubMed

See more...

RefSNP page

Reference SNP (refSNP) Cluster Report: rs8193049

RefSNP	Allele	Links
Organism: cow (Bos taurus)	Variation Class : SNV: single nucleotide variation	
Molecule Type: Genomic	RefSNP Alleles: A/C (FWD)	
Created/Updated in build: 117/142	Allele Origin:	
Map to Genome Build: 103/Weight	Ancestral Allele: Not available	
Validation Status :	MAF/MinorAlleleCount: NA	
	MAF Source:	

SNP Details are organized in the following sections:

[GeneView](#)
[Map](#)
[Submission](#)
[Fasta](#)
[Resource](#)
[Diversity](#)
[Validation](#)

Integrated Maps (Hint: click on 'Chr Pos' to see variant in the new NCBI variation viewer)



Assembly	Annotation Release	Chr	Chr Pos	Contig	Contig Pos	SNP to Chr	Contig allele	Contig to Chr	Neighbor SNP	Map Method
Bos_taurus_UMD_3.1	103	8	108837116	NW_003104150.1	108031	Fwd	A	Fwd	view	remap
Btau_4.6.1	103	8	112334706	NW_001495497.3	3734334	Fwd	A	Fwd	view	remap

Variant in gene context

GeneView ↑

GeneView via analysis of contig annotation: [TLR4](#) toll-like receptor 4

▼ View more variation on this gene (click to hide).

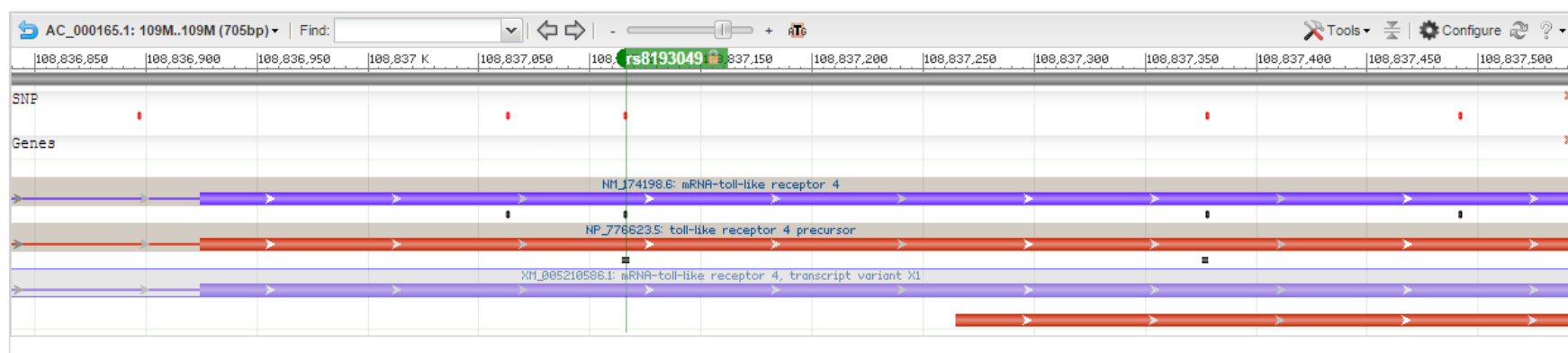
☐ in gene region ☒ cSNP ☐ has frequency ☐ double hit

Primary Assembly Mapping

Assembly	SNP to Chr	Chr	Chr position	Contig	Contig position	Allele
Bos_taurus_UMD_3.1	Fwd	8	108837116	NW_003104150.1	108031	A

Gene Model(s)














Function	mRNA				Protein		
	SNP to mRNA	Accession	Position	Allele change	Accession	Position	Residue change
missense	Fwd	NM_174198.6	922	AAC ⇒ ACC	NP_776623.5	151	N [Asn] ⇒ T [Thr]



dbSNP FTP

<ftp://ftp.ncbi.nih.gov/snp/organisms/>

Index of /snp/organisms/cow_9913/

Name	Size	Date Modified
 [parent directory]		
 ASN1_bin/		3/30/14, 12:00:00 AM
 ASN1_flat/		3/30/14, 12:00:00 AM
 BED/		3/30/14, 12:00:00 AM
 VCF/		3/30/14, 12:00:00 AM
 XML/		3/30/14, 12:00:00 AM
 chr_rpts/		3/30/14, 12:00:00 AM
 database/		2/13/06, 12:00:00 AM
 genotype/		9/20/12, 12:00:00 AM
 genotype_by_gene/		3/23/11, 12:00:00 AM
 rs_fasta/		3/30/14, 12:00:00 AM
 ss_fasta/		3/23/11, 12:00:00 AM
 submit_format/		10/30/05, 11:00:00 PM
 viewBatch/		7/17/14, 3:04:00 PM



dbVar – Primary archive for common and rare structural variation (somatic and germline)

<http://www.ncbi.nlm.nih.gov/dbvar>



dbVar

Database of genomic structural variation

Using dbVar

[Study Browser](#)

[Genome Browser](#)

[Download Data \(FTP\)](#)

[Overview of Structural Variation](#)

[FAQ](#)

[Help](#)

[Factsheet](#)

[dbVar News](#)

Submitting Data

[Submission Guidelines](#)

[Submission Templates](#)

[NEW! Submit Variants in VCF](#)

Other Resources

[NCBI Variation Portal](#)

[dbSNP](#)

[Database of Genomic Variants archive \(DGVa\)](#)

[Database of Genomic Variants \(DGV\)](#)

[NHGRI Structural Variation Project](#)

[ClinVar](#)

Allele Type

Depiction

Gain



Loss



Insertion



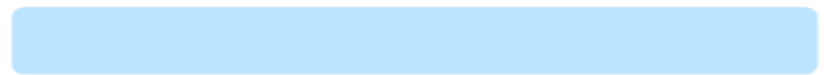
Tandem duplication



Inversion



Complex



UPD



Translocation



Unknown





Content dbVar

16 Organisms
127 Studies
1.3 Million Variations

TAXID	ORGANISM	VARIATION COUNT
4558	Sorghum bicolor	32261
7227	Drosophila melanogaster	70711
7955	Danio rerio	31749
9601	Pongo abelii	1329329
9612	Canis lupus	4149
9796	Equus caballus	193625
9823	Sus scrofa	570
9913	Bos taurus	10581
10090	Mus musculus	5001638
499232	Gorilla beringei	1329329
	more....	


Searching

Search using Genome Browser

dbVar Genome Browser **Bos taurus: Btau_4.6.1 (GCF_000003205.5)** [Reset All](#) [Share this page](#) [Help](#) [Back to Setup Page](#)

View data across studies on the genome


Select an Organism: **Bos taurus** Select an Assembly: **Btau_4.6.1**


Q- BPIFA2A 

Enter a location, gene name or phenotype
► Search examples:

Search using Entrez

NCBI Resources How To [Sign in to NCBI](#)

dbVar **dbVar** **PTEN**  **Search** [Save search](#) [Limits](#) [Advanced](#) [Help](#)


Display Settings: ☒ Tabular View, 20 per page **Send to:** 


Results: 1 to 20 of 44 [<< First](#) [< Prev](#) Page **1** of 3 [Next >](#) [Last >>](#)

Studies (44) **Variants (222)**


Number of Studies: 20

Study ID	Organism	Study type	Number of variant regions	Publication
estd214	human	Control Set	62855	
nstd101	human	Case-Set	3700	Kaminsky et al. 2011
estd212	human	Control Set	16575	
nstd100	human	Case-Control	70319	Coe et al. 2014
estd215	human	Control Set	28083	Boomsma et al. 2014
estd205	fruit fly	Control Set	12256	Zichner et al. 2013
nstd94	human	Tumor vs. Matched-Normal	8568	Helman et al. 2014
nstd92	human	Control Set	245	Forsberg et al. 2014

Find related data 


Database: **Select** 

[Find items](#)

Search details 

PTEN[All Fields]

[Search](#) [See more...](#)

Recent activity 

[Turn Off](#) [Clear](#)

Your browsing activity is empty.

dbVar study page

dbVar: Study Browser

Date	Publication	Study	Organism	Variant Region Count	Variant Call Count
2014/10	1000 Genomes Consortium Phase 3	estd214	Human	62,855	6,623,477
2014/10	Sallustio et al. 2014	nstd91	Human	148	148
2014/09	Uddin et al. 2014	estd212	Human	16,575	71,178
2014/09	Coe et al. 2014	nstd100	Human	70,319	318,775
2014/09	Campbell et al. 2014	nstd98	Human	16	18
2014/07	Campbell et al. 2014	estd211	Human	100	100
2014/06	Forsberg et al. 2014	nstd92	Human	245	245
2014/05	Helman et al. 2014	nstd94	Human	8,568	177,458
2014/05	Lindstrand et al. 2014	nstd93	Human	1	1
2014/02	Boomsma et al. 2014	estd215	Human	28,083	28,083
2014/01	de Boer et al. 2014	nstd89	Human	1	1
2014/01	Dogan et al. 2014	nstd73	Human	9,109	9,109
2013/12	Vulto-van Silfhout et al. 2013	nstd85	Human	1,416	1,663
2013/11	Pang et al. 2013	estd209	Human	471,817	471,817
2013/11	Helbig et al. 2013	estd208	Human	81	88
2013/10	Poultney et al. 2013	nstd86	Human	1,030	1,386
2013/10	de Ligt et al. 2013	nstd84	Human	10,376	11,064
2013/09	Morak et al. 2013	nstd83	Human	25	35
2013/09	Dittwald et al. 2013	nstd79	Human	64	2,213
2013/07	Simon et al. 2013	estd204	Mouse	43	43
2013/07	Sudmant et al. 2013	nstd82	Bornean orangutan, Chimpanzee, Eastern gorilla, Human, Pygmy chimpanzee, Sumatran orangutan, Western gorilla	28,214	1,329,329
2013/05	Boone et al. 2013	nstd80	Human	1,512	3,785
2013/04	Chia et al. 2013	estd198	Human	381	399
2013/03	Zichner et al. 2013	estd205	Fruit fly	12,256	65,487
2013/03	Watson et al. 2013	nstd76	Human	9	12
2013/03	Kazmi et al. 2013	nstd74	Mouse	172	172
2013/01	Wong et al. 2013	estd201	Human	36,558	312,665
2013/01	Schrider et al. 2013	nstd78	Human	38	38
2012/12	Wapner et al. 2012	nstd75	Human	925	2,417

Filter by Organism

Human (99)

Mouse (12)

Cattle (4)

Chimpanzee (3)

Fruit fly (3)

See more...
Source: NCBI

Filter by Study Type

Control Set (92)

Case-Set (21)

Case-Control (9)

Tumor vs. Matched-Normal (3)

Curated Collection (2)

Source: NCBI

Filter by Method

Oligo aCGH (24)

Sequencing (24)

SNP array (16)

BAC aCGH (7)

SNP genotyping analysis (5)

See more...
Source: NCBI

Filter by Variant

>=10

>=100

>=1,000

>=10,000

>=100,000

Source: NCBI

dbVar study page

nstd69

Organism: [Cattle](#)

Study Type: [Control Set](#)

Submitter: [Derek Bickhart](#)

Description: Using a read depth approach based on next-generation sequencing, we examined genome-wide copy number differences among five taurine (three Angus, one Holstein and one Hereford) and one indicine (Nelore) cattle. We identified 1,265 CNV regions comprising ~55.6 Mbp sequence-476 of which (~38%) have not previously been reported. Genes related to pathogen- and parasite-resistance, such as CATHL4 and ULBP17, were highly duplicated in the Nelore individual relative to the taurine cattle, while genes involved in lipid transport and metabolism, including APOL3 and FABP2, were highly duplicated in the beef breeds. These CNV regions also harbor genes like BPIFA2A (BSP30A) and WC1, suggesting that some CNVs may be associated with breed-specific differences in adaptation, health, and production traits.

Publication(s): [Bickhart et al. 2012](#)

Links

[Variants in this study](#)

[Open Cattle in Taxonomy Browser](#)

Source: NCBI

Detailed Information: [Download 1265 Variant Regions](#), [Download 4697 Variant Calls](#), [Download Both](#), [FTP](#)

Variant Summary

Samplesets

Experimental Details

Validations

Assembly used for analysis:
Remapped: Btau_4.6.1
Submitted: Btau_4.0 (bosTau4)
Remapped: Bos_taurus_UMD_3.1

Variant Summary for: Btau_4.6.1

Items 1 - 10 of 32

<< First

< Prev

Page 1 of 4

Next >

Last >>

Sequence ID	Chr	Number of Variant Regions	Number of Variant Calls	Placement type	Link to graphical display
NC_007299.5	Chr1	41	135	Remapped	NC_007299.5
NC_007300.5	Chr2	38	112	Remapped	NC_007300.5
NC_007301.5	Chr3	72	309	Remapped	NC_007301.5
NC_007302.5	Chr4	49	185	Remapped	NC_007302.5
NC_007303.5	Chr5	71	306	Remapped	NC_007303.5
NC_007304.5	Chr6	34	117	Remapped	NC_007304.5
NC_007305.5	Chr7	65	211	Remapped	NC_007305.5
NC_007306.5	Chr8	45	180	Remapped	NC_007306.5
NC_007307.5	Chr9	26	108	Remapped	NC_007307.5
NC_007308.5	Chr10	43	171	Remapped	NC_007308.5

Items 1 - 10 of 32

<< First

< Prev

Page 1 of 4

Next >

Last >>

Remapping summary: Btau_4.0->Bos_taurus_UMD_3.1

dbVar genome browser

NCBI Resources How To lonphan My NCBI Sign Out

dbVar Genome Browser Bos taurus: Btau_4.6.1 (GCF_000003205.5) Chr 13 (NC_007311.5): 63.36M - 63.38M

Reset All Share this page Help Back to Setup Page

View on Genome

1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 X Y

Select Assembly

Btau_4.6.1
Select an assembly to change view

Search

Q- BPIFA2A
Enter a location, gene name or phenotype

Name	Location
NM_174803.3	Chr13 63.36M - 63.38M

Your Data

Region Summary

Data in view Click (-) to remove track

Study ID	Variant Calls
nstd69	6 (-)

Data available for region Click (+) to add track

Study ID	Variant Calls
nstd56	11 (+)

Click (+) to add the track. Click (-) to remove the track.
You can add/remove multiple tracks at a time.

NC_007311.5: 63M..63M (24Kbp)

63,358 K 63,360 K 63,362 K 63,364 K 63,366 K 63,368 K 63,370 K 63,372 K 63,374 K 63,376 K 63,378 K 63,380 K

Genes

NM_174803.3 BPIFA2A NP_777228.2

Gnomon Alignments

Refseq Alignments

dbVar Sickhart et al. 2012 (nstd69)

dbVar variant report page

nsv835143

Organism: [Bos taurus](#)

Study: [nstd69 \(Bickhart et al. 2012\)](#)

Variant Type: copy number variation

Method Type: Sequencing

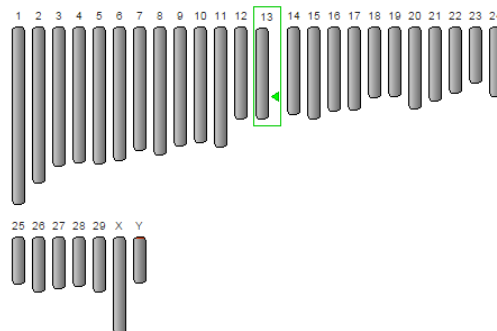
Submitted on: Btau_4.0 (bosTau4)

Publication(s): [Bickhart et al. 2012](#)

Variant Calls: 6

Validation: Not tested

Region Size: 42,467



[Links to Other Resources](#)

[Overlapping Genes](#)

Source: NCBI

Btau_4.6.1

◀ nsv835143

Genome View

Variant Region Details and Evidence

Validation Information

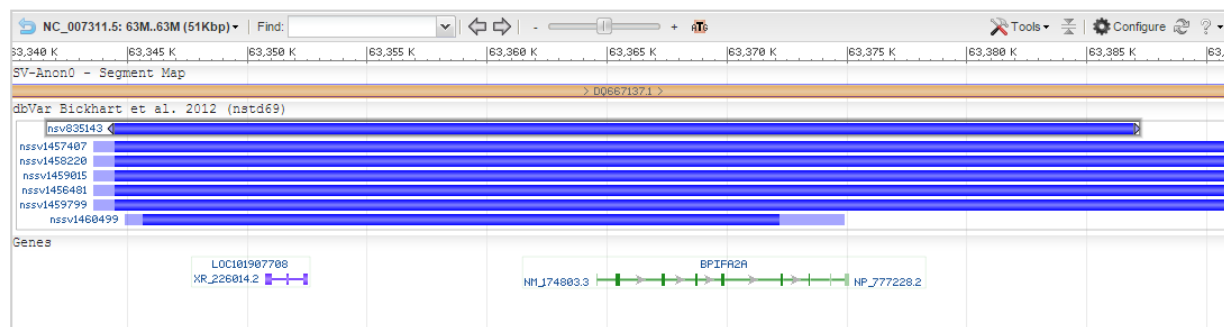
Genotype Information

Genome View

Select assembly: [Btau_4.6.1: Chr13](#)

Overlapping variant regions from other studies: 1 SVs from 1 studies. See in: [genome view](#)

Remapped (Score: Pass): [63,344,468](#) - [63,386,934](#)



ClinVar

ClinVar

Search ClinVar for gene symbols, HGVS expressions, conditions, and more

Search

Advanced

Help

Home

About

Data use and maintenance

Using the website

How to submit

Statistics

FTP site

ACTGATGGTATGGGGCCAAGAGATATATCT
 CAGGTACGGCTGTCATCACTTAGACCTCAC
 CAGGGCTGGGCATAAAAGTCAGGGCAGAGC
 CCATGGTGCATCTGACTCCTGAGGAGAAGT
 GCAGGTTGGTATCAAGGTTACAAGACAGGT
 GGCACCTGACTCTCTCTGCCTATTGGTCTAT

ClinVar

ClinVar aggregates information about sequence variation and its relationship to human health.

Using ClinVar

[About ClinVar](#)

[Data Dictionary](#)

[Downloads/FTP site](#)

[FAQ](#)

[Contact Us](#)

[RSS feed/What's new?](#)

[Factsheet](#)

Tools

[ACMG Recommendations for Reporting of Incidental Findings](#)

[Clinical Remapping - Between assemblies and RefSeqGenes](#)

[RefSeqGene/LRG](#)

[Submissions](#)

[Variation Reporter](#)

[Variation Viewer](#)

Related Sites

[ClinGen](#)

[GeneReviews®](#)

[GTR®](#)

[ICCG](#)

[MedGen](#)

[OMIM®](#)

[Variation](#)

Submitter highlights

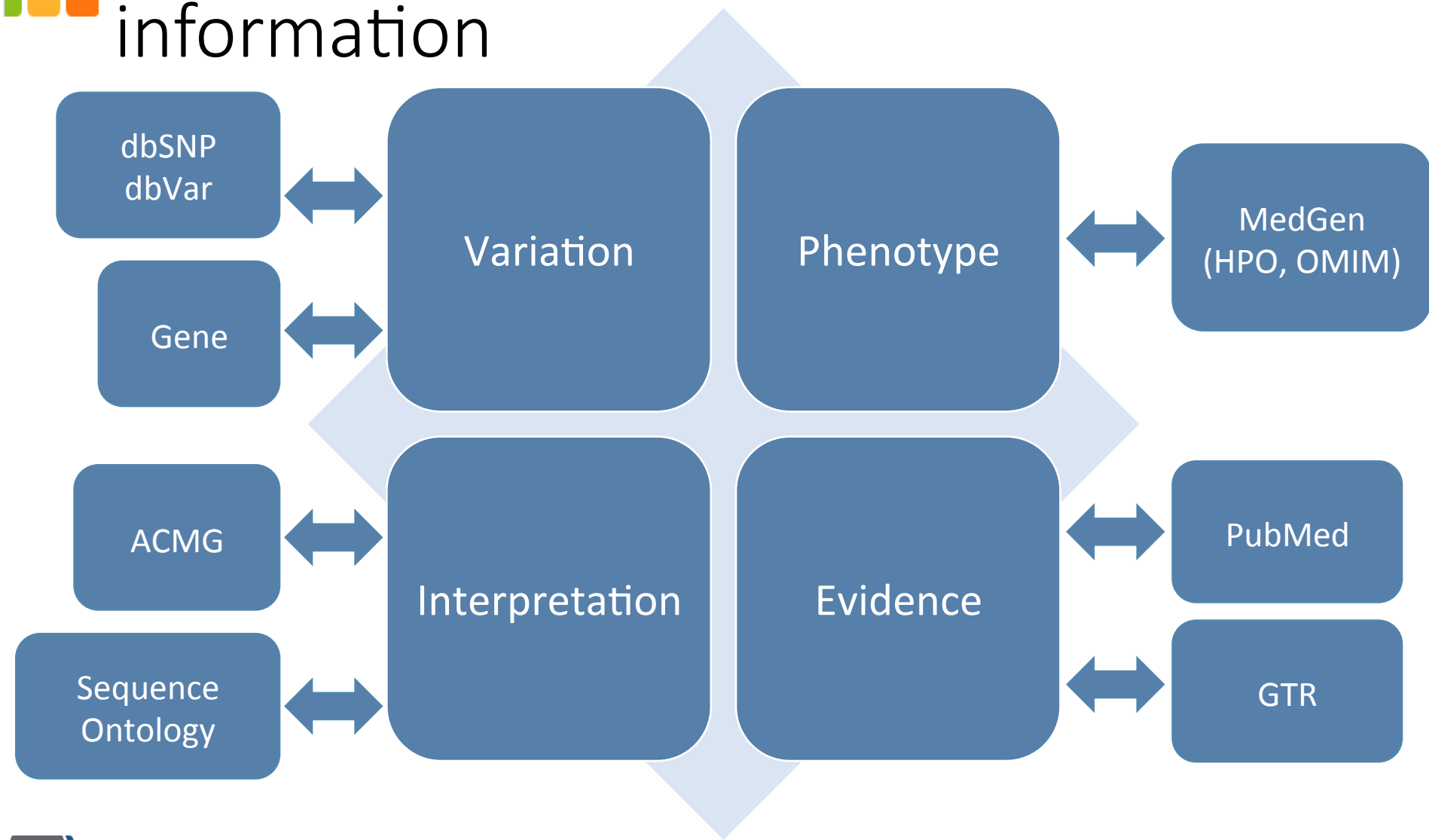
We gratefully acknowledge those who have submitted data and provided advice during the development of ClinVar. Subscribe to our [RSS feed](#) to receive announcements of the release of new datasets. More [information about our submitters](#) is available, as well as a list of submitters with [the number of records each has submitted](#).

Disclaimer

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. NIH does not independently verify the submitted information. If you have questions about the information contained on this website, please see a health care professional. More information about [NCBI's disclaimer policy](#) is available.



ClinVar aggregates four domains of information



Gene

Customize this list...

Clinical
significanceConflicting
interpretations (16)

Benign (37)

Likely benign (7)

Uncertain significance (20)

Likely pathogenic (0)

Pathogenic (13)

Risk factor (3)

Review status

Professional society (0)

Expert panel (0)

Multiple submitters (30)

Single submitter (26)

Method type

Research (8)

Literature only (28)

Clinical testing (56)

Molecular

consequence

Frameshift (3)

Missense (13)

Nonsense (2)

Splice site (0)

ncRNA (16)

Near gene (0)

UTR (1)

Variation type

Copy gain (0)

Copy loss (0)

Deletion (12)

Duplication (3)

indel (0)

Insertion (3)

Single nucleotide (47)

Display Settings: ☒ Tabular, 20 per page, Sorted by Default orderSend to: ☐

Results: 1 to 20 of 62

<< First < Prev Page 1 of 4 Next > Last >>

	Gene(s)	Condition(s)	Frequency	Clinical significance (Last reviewed)	Review status	Chr	Location (GRCh38)
1. <input type="checkbox"/>	NM_000059.3(BRCA2):c.5555T>A (p.Val1852Asp)	BRCA2	Familial cancer of breast	not provided	not classified by submitter	13	32339910
2. <input type="checkbox"/>	NM_007294.3(BRCA1):c.5074+284C>A	BRCA1	Familial cancer of breast	GMAF:0.25280(T)	not provided	not classified by submitter	17 43067324
3. <input type="checkbox"/>	NM_007294.3(BRCA1):c.5074+65G>A	BRCA1	Familial cancer of breast	GMAF:0.30690(T)	not provided	not classified by submitter	17 43067543
4. <input type="checkbox"/>	NM_007294.3:c.671-248_671-246dupAGG	BRCA1	Familial cancer of breast	not provided	not classified by submitter	17	43095106 - 43095108
5. <input type="checkbox"/>	NM_000059.3:c.6841+80_6841+83delTTAA	BRCA2	Familial cancer of breast, Breast- ovarian cancer, familial 2	Uncertain significance (Dec 17, 2010)	classified by single submitter	13	32341276 - 32341279
6. <input type="checkbox"/>	NM_007294.3(BRCA1):c.4097-141A>C	BRCA1	Familial cancer of breast, Breast- ovarian cancer, familial 1	GMAF:0.49760(T)	Uncertain significance (Aug 7, 2009)	classified by single submitter	17 43091173
7. <input type="checkbox"/>	NM_000059.3(BRCA2):c.-26G>A	BRCA2	Familial cancer of breast, Breast- ovarian cancer, familial 2	GO-ESP:0.20883(A) GMAF:0.20930(A)	Benign/Likely benign (Mar 17, 2011)	classified by multiple submitters	13 32316435
8. <input type="checkbox"/>	NM_007294.3(BRCA1):c.2216_2217delAA (p.Lys739Serfs)	BRCA1	Familial cancer of breast	not provided (Feb 1, 2013)	not classified	17	43093314 -

Variant summary

- Links to dbSNP
- Links to browsers including variation viewer
- Summary of aggregated information about the variant as well as details per submitter.
- Highlights clinical significance and review status.

ClinVar Search ClinVar for gene symbols, HGVS expressions, conditions, and more [Advanced](#) [Help](#)

Home About Data use and maintenance Using the website How to submit Statistics FTP site

NM_000059.3(BRCA2):c.3396A>G (p.Lys1132=)

NM_000059.3(BRCA2):c.3396A>G (p.Lys1132=)

Go to:

Clinical significance
 NM_000059.3(BRCA2):c.3396A>G (p.Lys1132=) [Help](#)
 Clinical significance: [Benign/Likely benign](#)
 Review status: ★ ★ ★ ★
 Number of submission(s): 6

Condition(s)
 Familial cancer of breast [\[MedGen - OMIM\]](#)
 Breast-ovarian cancer, familial 2 [\[MedGen - Orphanet - OMIM\]](#)
 Neoplastic Syndromes, Hereditary [\[MedGen\]](#)

[See supporting ClinVar records](#)

1 Affected Gene

breast cancer 2, early onset (BRCA2) [\[Gene - OMIM - Variation viewer\]](#)
 Haploinsufficiency - Sufficient evidence for dosage pathogenicity (Jul 6, 2012)
 Triplosensitivity - No evidence available (Jul 6, 2012)
 🔍 Search ClinVar for variants within BRCA2
 🔍 Search ClinVar for variants including BRCA2

Browser views

RefSeqGene
 Variation viewer [\[GRCh38 - GRCh37\]](#)
 UCSC [\[GRCh38/hg38 - GRCh37/hg19\]](#)

Related information

dbSNP
 Gene
 MedGen
 OMIM

Recent Activity

Assertion and evidence details [Go to:](#)

[Clinical Assertions](#) [Evidence](#) [Help](#)

Germline

Clinical significance (Last evaluated)	Review status (Assertion method)	Collection method	Condition(s) (Mode of inheritance)	Origin	Citations	Submitter (Last submitted)	Submission accession
Benign (Feb 20, 2004)	classified by single submitter (clinical testing)	clinical testing	Breast-ovarian cancer, familial 2 [MedGen Orphanet OMIM]	germline		Breast Cancer Information Core (BIC) (BRCA2) (Mar 28, 2014)	SCV000146225
Benign (Jan 2, 2014)	classified by single submitter (literature only)	literature only	Breast-ovarian cancer, familial 2 (Autosomal dominant inheritance) [MedGen Orphanet OMIM]	unknown		Counsyl (May 1, 2014)	SCV000154055
Benign (Sep 4, 2013)	classified by single submitter (clinical testing)	clinical testing	not provided [MedGen]	germline		GeneDx (Jun 10, 2014)	SCV000167353
Benign (Jul 10, 2012)	classified by single submitter (clinical testing)	clinical testing	Neoplastic Syndromes, Hereditary [MedGen]	germline		Ambry Genetics (Jul 25, 2014)	SCV000185907

Searching by genomic location

- Variation Viewer

Variation Viewer

Homo sapiens: GRCh38 (GCF_000001405.26) Chr 2 (NC_000002.12): 41.50M - 54.70M

Reset All Share this page FAQ Help Version 1.1.3

Pick Assembly

Search

2p21-2p16.2

Enter a location, gene name or phenotype

Genes Other features

Name	Location
2p21-2p16.2	Chr2 41.50M - 54.70M

Your Data

History

Region Details

Features of Interest

Other sequence representations - None

No GRC issues in this view [Add Track](#)

Exon Navigator

There are too many genes in the region (124). Please narrow the region to enable exon navigation.

NC_000002.12: 41M..55M (13Mbp)

42 M 43 M 44 M 45 M 46 M 47 M 48 M 49 M 50 M 51 M 52 M 53 M 54 M

Genes, NCBI Homo sapiens Annotation Release 106

PRKCE EPAS1 EPCAM MSH2 MSH6 LHCGR FSHR

ClinVar Short Variations based on dbSNP 141 (Homo sapiens Annotation Release 106)

dbSNP 141 (Homo sapiens Annotation Release 106) all data

ClinVar Large Variations based on dbVar

Variation Data

Filter by

Source database

☐ dbSNP (1,071)

☐ dbVar (23)

In ClinVar

☒ Yes (1,094)

☐ No (0)

Worst clinical significance

☐ Pathogenic (476)

☐ Likely pathogenic (57)

☐ Benign (1)

Download Items 1 - 20 of 1,094 << First < Prev Page 1 of 55 Next > Last >>

Variant ID	Location	Variant type	Gene	Molecular consequences	Worst clinical significance	1000G MAF	GO-ESP MAF	Publications
ns531646	66,097 - 55,570,637	copy number variation	TSPYL6 and 347 more		Pathogenic			2

Variant calls associated with ns531646

Allele information		ClinVar information		Worst clinical significance	Submitters	Highest review status	Last evaluated
Call ID	Change	Description					
nssv578821	copy number gain	Developmental delay and additional significant developmental and morphological phenotypes referred for genetic testing		Pathogenic	1	classified by single submitter	Aug 12, 2011



Variation reporter

<http://www.ncbi.nlm.nih.gov/variation/tools/reporter/>

- Provides access to information about sequence variation at different locations on a genome
- Useful for a few variants or a large batch
- Accepts GVF, VCF, BED, HGVS formats
- Returns both a sample visual display and files for download
- Interactive or API

NCBI genome remapping service

NCBI Genome Remapping Service

* indicates required fields.

Assembly-Assembly Clinical Remap Alt loci remap

Genome Information

Source Organism *

Bos taurus

Start typing to get a list of available organisms

Select the assembly on which your data is annotated.
Use the [Write to the Help Desk](#) to request the addition of an assembly that is not in the list.

Source Assembly *

Bos_taurus_UMD_3.1.1
Bos_taurus_UMD_3.1
Btau_4.6.1 (bosTau7)
Btau_4.2

Target Assembly *

Remapping Options

Minimum ratio of bases that must be remapped: 0.5

Maximum ratio for difference between source length and target length: 2.0

Allow multiple locations to be returned: ☒

Merge Fragments: ☒

NCBI Remap

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[About our alignments](#)

[FAQ](#)

[API Documentation](#)

[Alignment FTP](#)

[About Genome Workbench](#)

[Write to the Help Desk](#)

Data

Input format: Best Guess Output format: Same as input

Upload a file: Browse... No file selected.

OR

Paste data here:

NCBI Genome Resources Workshop

- 12:50– 1:15 Improving the Flow of Data to NCBI Sequence Repositories, SRA and GenBank
Ilene Mizrachi
- 1:15– 1:40 Variation at NCBI: Resources, Tools and Submissions
Jennifer Lee
- 1:40– 2:05 Update on the Eukaryotic Genome Annotation Pipeline
Francoise Thibaud-Nissen
- 2:05– 2:30 From Genes to Genomes – New Features and Data Access
Kim D. Pruitt
- 2:30– 2:50 Annual Report on Genome Sequencing Projects
Tatiana Tatusova
- 2:50-3:00 Questions & Discussion

Poster 1109 – dbSNP and dbVar

NCBI Booth 618
info@ncbi.nlm.nih.gov

